



dermatofibrosarcoma protuberans

Dermatofibrosarcoma protuberans is a rare type of cancer that causes a tumor in the deep layers of skin. This condition is a type of soft tissue sarcoma, which are cancers that affect skin, fat, muscle, and similar tissues.

In dermatofibrosarcoma protuberans, the tumor most often starts as a small, firm patch of skin, usually 1 to 5 centimeters in diameter, that is usually purplish, reddish, or flesh-colored. The tumor typically grows slowly and can become a raised nodule. Occasionally, the tumor begins as a flat or depressed patch of skin (plaque). Tumors are most commonly found on the torso and can also be found on the arms, legs, head, or neck. Affected individuals usually first show signs of this condition in their thirties, but the age at which a tumor appears varies widely.

In dermatofibrosarcoma protuberans, the tumor has a tendency to return after being removed. However, it does not often spread to other parts of the body (metastasize).

There are several variants of dermatofibrosarcoma protuberans in which different cell types are involved in the tumor. Bednar tumors, often called pigmented dermatofibrosarcoma protuberans, contain dark-colored (pigmented) cells called melanin-containing dendritic cells. Myxoid dermatofibrosarcoma protuberans tumors contain an abnormal type of connective tissue known as myxoid stroma. Giant cell fibroblastoma, which is sometimes referred to as juvenile dermatofibrosarcoma protuberans because it typically affects children and adolescents, is characterized by giant cells in the tumor.

Rarely, the tumors involved in the different types of dermatofibrosarcoma protuberans can have regions that look similar to fibrosarcoma, a more aggressive type of soft tissue sarcoma. In these cases, the condition is called fibrosarcomatous dermatofibrosarcoma protuberans or FS-DFSP. FS-DFSP tumors are more likely to metastasize than tumors in the other types of dermatofibrosarcoma protuberans.

Frequency

Dermatofibrosarcoma protuberans is estimated to occur in 1 in 100,000 to 1 in 1 million people per year.

Genetic Changes

Dermatofibrosarcoma protuberans is associated with a rearrangement (translocation) of genetic material between chromosomes 17 and 22. This translocation, written as t(17;22), fuses part of the *COL1A1* gene from chromosome 17 with part of the *PDGFB* gene from chromosome 22. The translocation is found on one or more

extra chromosomes that can be either the normal linear shape or circular. When circular, the extra chromosomes are known as supernumerary ring chromosomes. Ring chromosomes occur when a chromosome breaks in two places and the ends of the chromosome arms fuse together to form a circular structure. Other genes from chromosomes 17 and 22 can be found on the extra chromosomes, but the role these genes play in development of the condition is unclear. The translocation is acquired during a person's lifetime and the chromosomes containing the translocation are present only in the tumor cells. This type of genetic change is called a somatic mutation.

In normal cells, the *COL1A1* gene provides instructions for making part of a large molecule called type I collagen, which strengthens and supports many tissues in the body. The *PDGFB* gene provides instructions for making one version (isoform) of the platelet derived growth factor (PDGF) protein. By attaching to its receptor, the active PDGFB protein stimulates many cellular processes, including cell growth and division (proliferation) and maturation (differentiation).

The abnormally fused *COL1A1-PDGFB* gene provides instructions for making an abnormal combined (fusion) protein that researchers believe ultimately functions like the PDGFB protein. The gene fusion leads to the production of an excessive amount of protein that functions like the PDGFB protein. In excess, this fusion protein stimulates cells to proliferate and differentiate abnormally, leading to the tumor formation seen in dermatofibrosarcoma protuberans.

The *COL1A1-PDGFB* fusion gene is found in more than 90 percent of dermatofibrosarcoma protuberans cases. In the remaining cases, changes in other genes may be associated with this condition. These genes have not been identified.

Inheritance Pattern

Dermatofibrosarcoma protuberans results from a new mutation that occurs in the body's cells after conception and is found only in the tumor cells. This type of genetic change is called a somatic mutation and is generally not inherited.

Other Names for This Condition

- Darier-Ferrand tumor
- Darier-Hoffmann tumor
- dermatofibrosarcoma
- DFSP

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Dermatofibrosarcoma protuberans
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0392784/>

Other Diagnosis and Management Resources

- American Cancer Society: How are Soft Tissue Sarcomas Diagnosed?
<https://www.cancer.org/cancer/soft-tissue-sarcoma/detection-diagnosis-staging/how-diagnosed.html>
- American Cancer Society: Treatment of Soft Tissue Sarcomas
<https://www.cancer.org/cancer/soft-tissue-sarcoma/treating/by-stage.html>
- National Cancer Institute: Adult Soft Tissue Sarcoma
<https://www.cancer.gov/types/soft-tissue-sarcoma/patient/adult-soft-tissue-treatment-pdq>
- National Cancer Institute: Targeted Cancer Therapies
<https://www.cancer.gov/about-cancer/treatment/types/targeted-therapies/targeted-therapies-fact-sheet>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Health Topic: Soft Tissue Sarcoma
<https://medlineplus.gov/softtissuesarcoma.html>

Genetic and Rare Diseases Information Center

- Dermatofibrosarcoma protuberans
<https://rarediseases.info.nih.gov/diseases/9569/dermatofibrosarcoma-protuberans>

Additional NIH Resources

- National Cancer Institute: Adult Soft Tissue Sarcoma
<https://www.cancer.gov/types/soft-tissue-sarcoma/patient/adult-soft-tissue-treatment-pdq>
- National Cancer Institute: Childhood Soft Tissue Sarcoma
<https://www.cancer.gov/types/soft-tissue-sarcoma/patient/child-soft-tissue-treatment-pdq>
- National Cancer Institute: Targeted Cancer Therapies
<https://www.cancer.gov/about-cancer/treatment/types/targeted-therapies/targeted-therapies-fact-sheet>

Educational Resources

- American Academy of Dermatology Skin Cancer Net: Dermatofibrosarcoma Protuberans (DFSP)
<https://www.aad.org/public/diseases/skin-cancer/dermatofibrosarcoma-protuberans>
- American Cancer Society: What is a Soft Tissue Sarcoma?
<https://www.cancer.org/cancer/soft-tissue-sarcoma.html>
- Disease InfoSearch: Dermatofibrosarcoma Protuberans
<http://www.diseaseinfosearch.org/Dermatofibrosarcoma+Protuberans/2206>
- Liddy Shriver Sarcoma Initiative
<http://sarcomahelp.org/dermatofibrosarcoma-protuberans.html>
- MalaCards: dermatofibrosarcoma protuberans
http://www.malacards.org/card/dermatofibrosarcoma_protuberans
- MD Anderson Cancer Center: Soft Tissue Sarcoma
<https://www.mdanderson.org/cancer-types/soft-tissue-sarcoma/soft-tissue-sarcoma-facts.html>
- Orphanet: Dermatofibrosarcoma protuberans
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=31112

Patient Support and Advocacy Resources

- Cancer Care
<http://www.cancercare.org/>
- Sarcoma Foundation of America
<http://www.curesarcoma.org/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22dermatofibrosarcoma+protuberans%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Dermatofibrosarcoma%5BMAJR%5D%29+AND+%28dermatofibrosarcoma+protuberans%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

OMIM

- DERMATOFIBROSARCOMA PROTUBERANS
<http://omim.org/entry/607907>

Sources for This Summary

- Greco A, Fusetti L, Villa R, Sozzi G, Minoletti F, Mauri P, Pierotti MA. Transforming activity of the chimeric sequence formed by the fusion of collagen gene COL1A1 and the platelet derived growth factor b-chain gene in dermatofibrosarcoma protuberans. *Oncogene*. 1998 Sep 10;17(10):1313-9. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/9771975>
- Heldin CH, Ostman A, Rönstrand L. Signal transduction via platelet-derived growth factor receptors. *Biochim Biophys Acta*. 1998 Aug 19;1378(1):F79-113. Review. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/9739761>
- Kampshoff JL, Cogbill TH. Unusual skin tumors: Merkel cell carcinoma, eccrine carcinoma, glomus tumors, and dermatofibrosarcoma protuberans. *Surg Clin North Am*. 2009 Jun;89(3):727-38. doi: 10.1016/j.suc.2009.02.005. Review. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19465208>
- Mendenhall WM, Zlotecki RA, Scarborough MT. Dermatofibrosarcoma protuberans. *Cancer*. 2004 Dec 1;101(11):2503-8. Review. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15503305>
- Naeem R, Lux ML, Huang SF, Naber SP, Corson JM, Fletcher JA. Ring chromosomes in dermatofibrosarcoma protuberans are composed of interspersed sequences from chromosomes 17 and 22. *Am J Pathol*. 1995 Dec;147(6):1553-8. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/7495279>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1869963/>
- Pedeutour F, Simon MP, Minoletti F, Sozzi G, Pierotti MA, Hecht F, Turc-Carel C. Ring 22 chromosomes in dermatofibrosarcoma protuberans are low-level amplifiers of chromosome 17 and 22 sequences. *Cancer Res*. 1995 Jun 1;55(11):2400-3. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/7757993>
- Sandberg AA, Bridge JA. Updates on the cytogenetics and molecular genetics of bone and soft tissue tumors. Dermatofibrosarcoma protuberans and giant cell fibroblastoma. *Cancer Genet Cytogenet*. 2003 Jan 1;140(1):1-12. Review. *Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/12550751>

- Shimizu A, O'Brien KP, Sjöblom T, Pietras K, Buchdunger E, Collins VP, Heldin CH, Dumanski JP, Ostman A. The dermatofibrosarcoma protuberans-associated collagen type I α 1/platelet-derived growth factor (PDGF) B-chain fusion gene generates a transforming protein that is processed to functional PDGF-BB. *Cancer Res.* 1999 Aug 1;59(15):3719-23.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/10446987>
- Simon MP, Navarro M, Roux D, Pouyssegur J. Structural and functional analysis of a chimeric protein COL1A1-PDGFB generated by the translocation t(17;22)(q22;q13.1) in Dermatofibrosarcoma protuberans (DP). *Oncogene.* 2001 May 24;20(23):2965-75.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/11420709>
- Simon MP, Pedeutour F, Sirvent N, Grosgeorge J, Minoletti F, Coindre JM, Terrier-Lacombe MJ, Mandahl N, Craver RD, Blin N, Sozzi G, Turc-Carel C, O'Brien KP, Kedra D, Fransson I, Guilbaud C, Dumanski JP. Deregulation of the platelet-derived growth factor B-chain gene via fusion with collagen gene COL1A1 in dermatofibrosarcoma protuberans and giant-cell fibroblastoma. *Nat Genet.* 1997 Jan;15(1):95-8.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8988177>
- Sirvent N, Maire G, Pedeutour F. Genetics of dermatofibrosarcoma protuberans family of tumors: from ring chromosomes to tyrosine kinase inhibitor treatment. *Genes Chromosomes Cancer.* 2003 May;37(1):1-19. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12661001>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/dermatofibrosarcoma-protuberans>

Reviewed: September 2011

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications

U.S. National Library of Medicine

National Institutes of Health

Department of Health & Human Services